

PCT

WORLD INTELLECTUAL PROPERTY ORGANIZATION  
International Bureau



INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

<b>(51) International Patent Classification <sup>7</sup> :</b> <b>C12N 15/54, 9/10, C12Q 1/68, C07K</b> <b>16/40, G01N 33/50</b>		<b>A2</b>	<b>(11) International Publication Number:</b> <b>WO 00/05382</b> <b>(43) International Publication Date:</b> 3 February 2000 (03.02.00)
<b>(21) International Application Number:</b> PCT/IB99/01353 <b>(22) International Filing Date:</b> 23 July 1999 (23.07.99) <b>(30) Priority Data:</b> 60/093,940 23 July 1998 (23.07.98) US <b>(71) Applicant (for all designated States except US):</b> GENSET [FR/FR]; 24, rue Royale, F-75008 Paris (FR). <b>(72) Inventor; and</b> <b>(75) Inventor/Applicant (for US only):</b> BOUGUELERET, Lydie [FR/FR]; 14, rue Vouillé, F-75015 Paris (FR). <b>(74) Agents:</b> MARTIN, Jean-Jacques et al.; Cabinet Regimbeau, 26, avenue Kléber, F-75116 Paris (FR).			<b>(81) Designated States:</b> AE, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CU, CZ, DE, DK, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, UA, UG, US, UZ, VN, YU, ZA, ZW, ARIPO patent (GH, GM, KE, LS, MW, SD, SL, SZ, UG, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG).  <b>Published</b> <i>Without international search report and to be republished upon receipt of that report.</i>
<b>(54) Title:</b> A NUCLEIC ACID ENCODING A GERANYL-GERANYL PYROPHOSPHATE SYNTHETASE (GGPPS) AND POLYMORPHIC MARKERS ASSOCIATED WITH SAID NUCLEIC ACID			
<p>The diagram illustrates the structure of the nucleic acid sequence. It shows a genomic DNA sequence with markers at 1, 486, 732, 1376, and 1693. Below it, a cDNA sequence is shown with markers at 1, 115, 216, and 1414. The cDNA is labeled 'cDNA 1414b'. Below the cDNA, a CDS (Coding DNA Sequence) is shown with markers at 1, 115, 216, and 1414. Arrows indicate the mapping from genomic DNA to cDNA and from cDNA to CDS.</p>			
<b>(57) Abstract</b> <p>The present invention relates to a purified or isolated polynucleotide encoding human geranylgeranyl pyrophosphate synthetase, the regulatory nucleic acids contained therein, a polymorphic marker thereof and the resulting encoded protein, as well as to methods and kits for detecting this polynucleotide and this protein. The present invention also pertains to a polynucleotide carrying the natural regulatory regions of the <i>hGGPS</i> gene which is useful, for example, to express a heterologous nucleic acid in host cells or host organisms as well as functionally active regulatory polynucleotides derived from said regulatory region. The invention also consists in genetic markers, namely biallelic markers, which may be useful for the diagnosis of diseases related to an alteration in the regulatory or coding regions of <i>hGGPS</i>, such as pathologies related to a defect in the mevalonic biosynthetic pathway.</p>			